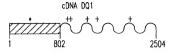
Laurie J. Ozelius et al. Inventors: 99A12 LL 60C12 LL 171B8 LL 158A7 LL COSMIDS 93H2 LL 54A5 LA 29A5 LA 23G1 LA 3D8 LA RECOMBINANT MARKER D9S63 9.8 X.4. 4.2 × × 9.7 × 9.8 × RECOMBINANT MARKER D9S2161 TRANSCRIPTS: × · × 9.7 171B8 LL 9312 11 COSMIDS 5445 LA 29A5 LA

Docket No.:

0838.1001-009 "Torsin, Torsin-Related Genes and Methods..." Docket No.: 0838.1001-009

"Torsin, Torsin-Related Genes and Methods ... "

Inventors: Laurie J. Ozelius et al.



Polymorphisms: C/T @ 343, proline/proline

Transcript size: 2.7kb

Transcript size: 2.2kb, 1.4kb



Polymorphisms: C/T @ 288, alanine/alanine

G/C @ 688, aspartic acid/histidine

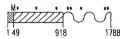
G/T @ 1232 C/G @ 1255

del/T @ 1464

T/Á @ 1495

Mutation: del/GAG @ 946, del/glutamic acid

cDNA DQ3



Polymorphisms: A/G @ 156, glutomic ocid/glutomic ocid A/G @ 420, lysine/lysine T/C @ 801, glycine/glycine

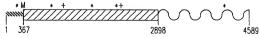
AC/CT @ 1005

G/A @ 1063

(Ť)n @ 1273 T/A @ 1724

A/G @ 1751

cDNA DQ4



Polymorphisms: G/A @ 225 C/T @ 840, alanine/alanine G/A @ 1696, voline/isoleucine

C/T @ 2172, histidine/histidine

G/A @ 4225

Transcript size: 4.5kb

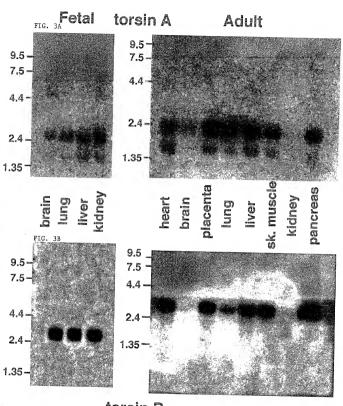
Transcript size: 1.8kb

FIG.2

Docket No.: 0838.1001-009

"Torsin, Torsin-Related Genes and Methods..."

Inventors: Laurie J. Ozelius et al.



DOTTE DE DIESTA

torsin B

0838.1001-009

"Torsin, Torsin-Related Genes and Methods Inventors: Laurie J. Ozelius et al.

NPKKPL TLSL HGVAGTGÅVEYSOI VAENL HPKGLKSNEVHLEVSTLHFPHEGK IKL YGDGLOKV IRGNVSACANSVF TFDEMDKLHPGI I DA IKPFLD-YY APSKPLVLSLHGVTG<u>TGK</u>SYVSSLLADALFRDGLRSPHVHHFSP11HFPHPSRTEQYKKELKSVVQGNLTACERSLFLFDEMDKLPPGLMEVLQPFLG-PS KPKKPLTLSLHGVTG<mark>TGR</mark>NFVSKTIAENIYEGGLNSDYVHLFVATLHFPHASNITLYKDQLQLVIRGNVSACARSIFIFDEMDKMHAGLIDATKPFLD-YY NPRKPL VLSFHGYTGSGÅYYVAET I ANNTFRLGLRSTF VQHTVATNJF PJKNKLEE YQVELRNRIL TTVQKCQRS IF IF DEAJKL PEQLLGATKPFLD-YY -----AAALHQTLF IFDEAEKLHPGLLEVLGPHLERRA - ATP-BINDING DOMAIN

> TorsinB TorsinA TorpCel

Torp1 Torp2

--MKLGRAVLGLLLAPSVVQAVEP ISLGLALAGVLTGY I YP------RLYCLFAECC-GOKRSLSREALOKDLDDNI.FGOHLAKK I ILMAVFGFINNP

TorsinA lorpCel OrsinB

orpi orp2

DOTTELLO DIVINE

MWMKL DY-VLLLLFHLCFVNTEL ISV ITGK IKDSGTTIA ISAGAF VGLK DRLKCYL YECCHEPDVNFNYHTL DADIANLL FGGHL VKDVVVNS IKSHWYNE

------LDLEEKLFGOHLATEVIFKALTGFRNNK

----LECDLAQHLAGQHLAKALVVKSLKAFVQDP

DLVDGVSYOKAMFIFLSNAGAERITDVALDFVRSGKOREDIKLKDIEHALSVSVFNNKNSGFWSSLIDPNLIDYFVPFLPLEYKHLKMCIRVEMOSRGY-EOVDGVSYXKA IF IFL SNAGGDLITKTALDF WRAGRKREDIOLKDLEPVL SVGVFNNKHSGL WHSGLIDKNLIDYF IPFL PLEYRHVKHCVRAEMRARGS-STISGVDFRRSIFILLSNKGGGEIAR ITKEDYESGYPREOLRLEAFERELMNFSYNEK-GGLOMSELISNHLIDHF VPFLPLOREHVRSCVGAYLRKRGRG OrsinB TorsinA lorpCel

PEXXGLSL XVT I FLFLSNLRGDI I NEVVLKLLKAGUSREE I TMEHLEPHLOAE I VDDHRDVLUHSRLVKENL I DYF I PFLPLEYRHVRLCARDAFL SQEL-

WVVYGTNYRKA IF IF I SNAGGE QI NDVALE AWRTNRDREE I SLOE VEPVI SRAVMDNPQHGF WRSG I MEEHLLDAVVPFLPLORHHVRHCVLNELAQLGL-

orp] orp2 --EIDEDIVSRVAEEMTFFPKEERVFSDKGCKTVFTKLDYYYDD --EPARRVFRRCVTD-TYFPEVEQLFSSNGCKTVASRLTFFL --AIDEDIVTRVAEEMTFFPRDEKIYSDKGCKTVQSRLDFH DL VSNV DF VERVLNSL QYF PESSKAF SSSGCKRV [orsin] orsinA lorpCel orpi

--LYKEETLDEIAQMMVYVPKEEQLFSSQGCKSIXQRIKLLPVMXG

orp2

FIG. 4A

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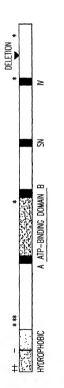


FIG. 4B